

# BIOLOGY

EST, SAT, ACT PREP



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## Chapter 4: Molecular Genetics

### First: Evidence

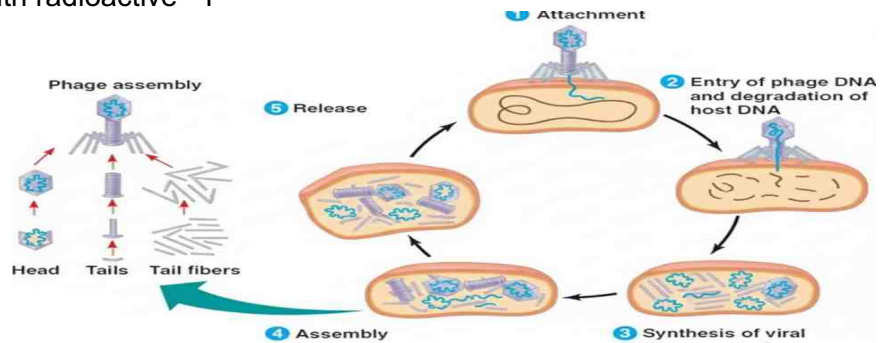
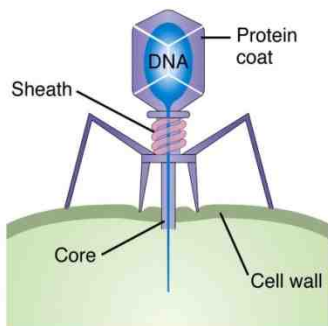
• **Bacterial transformation** **Griffith experiment** → Ability of bacteria (R) to alter their genetic makeup by absorbing foreign DNA molecules from other bacterial cells (S) and incorporating the foreign DNA into their own

Pneumonia Bacteria **R-strain**: non-virulent      **S-strain**: virulent

By injecting the bacteria into mice:

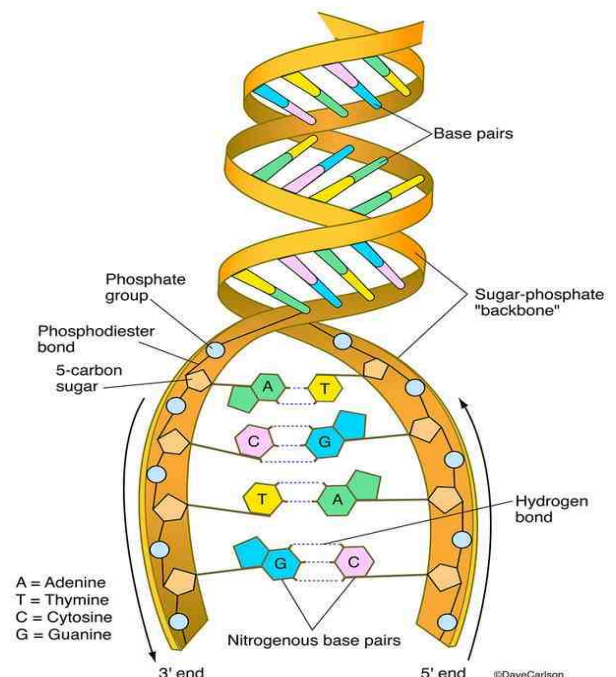
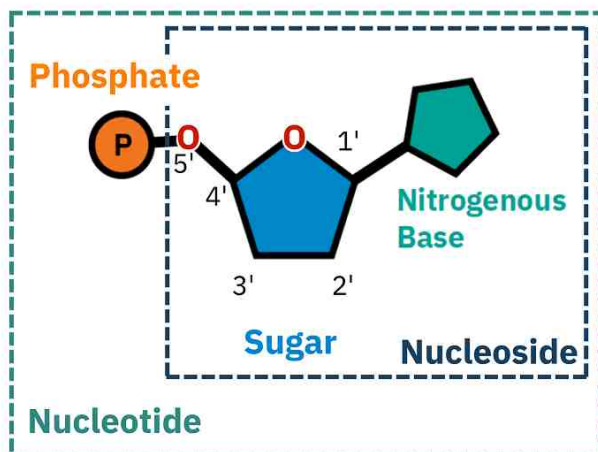
- R-strain → Survived
- S-strain → Died
- S-strain(heat killed) → Survived
- R + S (heat killed) → Died

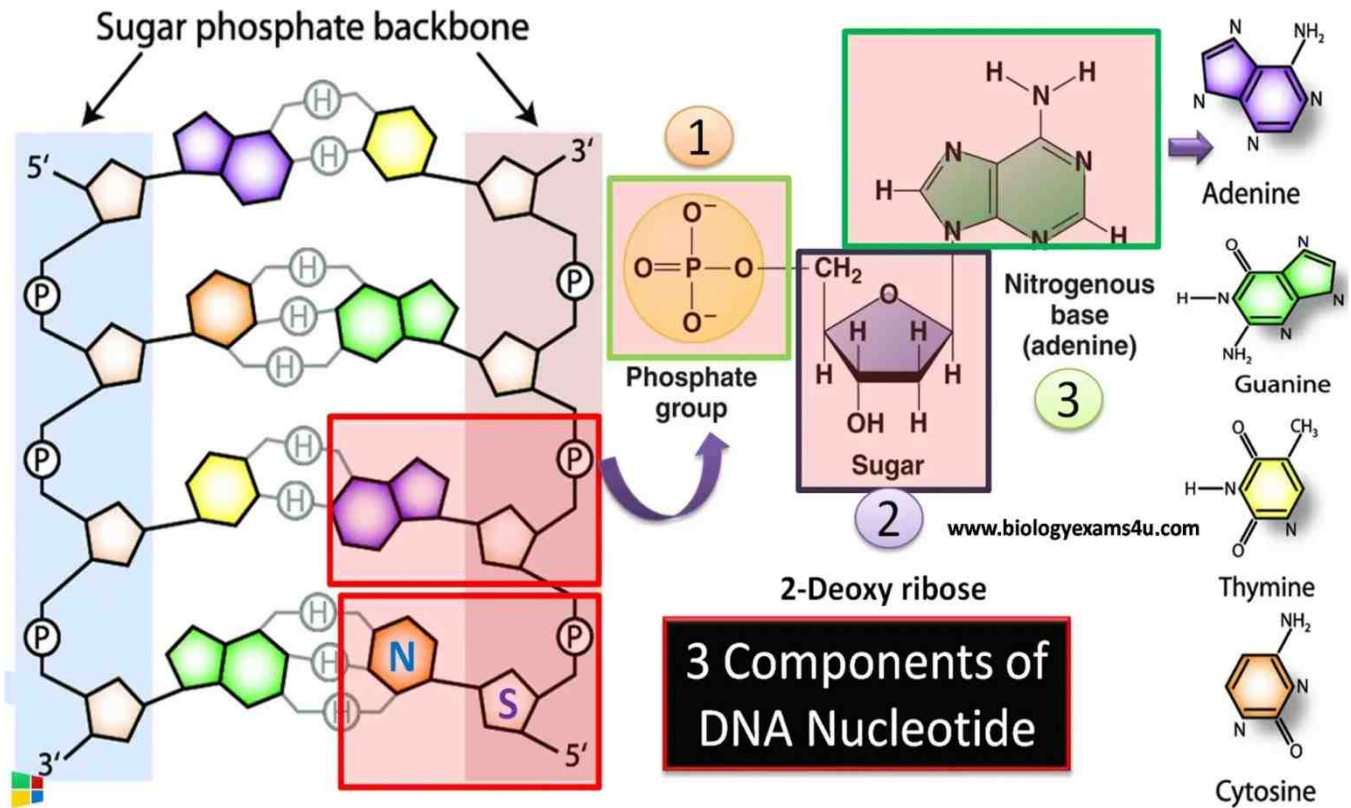
• **Bacteriophage** → virus attacking bacteria  
 • **Hershey and Chase (1952)** proved that DNA, not the protein, is the genetic material by labeling the bacteriophage (virus attacking bacteria), its DNA with radioactive  $^{32}\text{P}$  and its protein with radioactive  $^{35}\text{S}$



### Second: Structure of DNA

- **Nucleotide** → nitrogenous base, pentose & one phosphate
- **Base pairing rule**: A = T, G ≡ C (stronger)





## Structure of DNA

DNA is a polymer of nucleotides.

Chemical Components

- DNA: Deoxyribonucleic acid

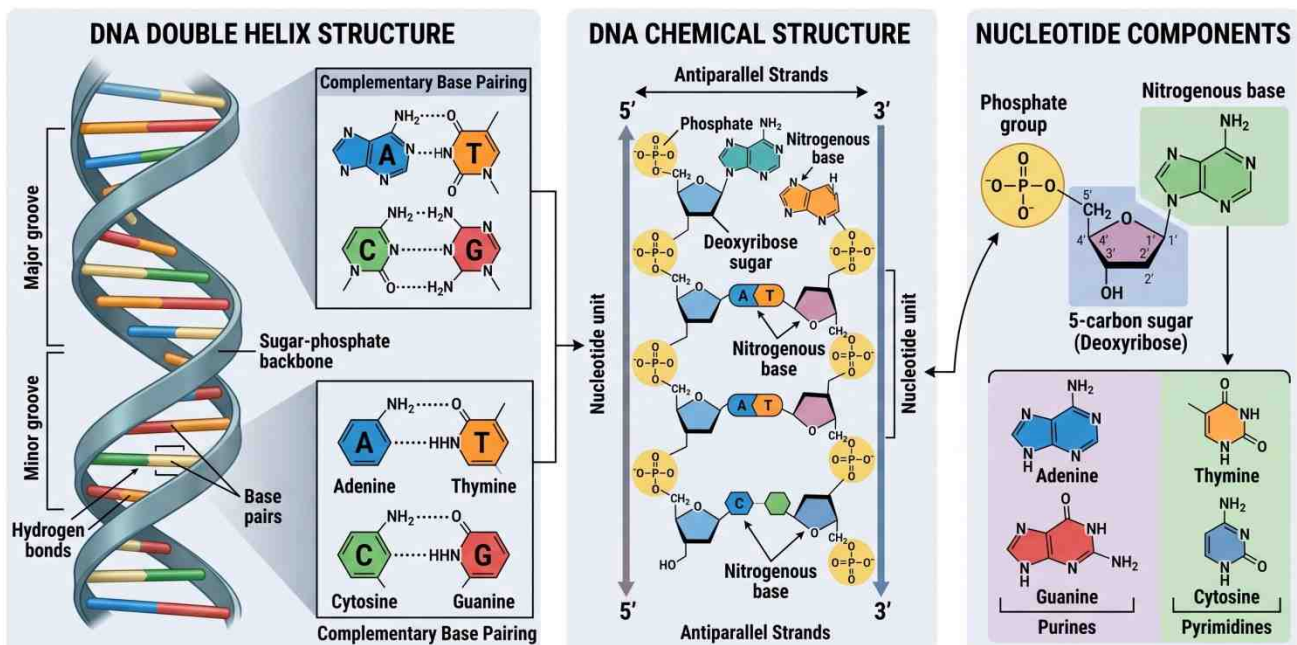
- RNA: Ribonucleic acid

Nitrogenous Bases

- Purine (double ring): Adenine (A) and Guanine (G)

- Pyrimidine (single ring): Thymine (T) and Cytosine (C)

# DIAGRAM FOR DNA STRUCTURE AND NUCLEOTIDE



**Franklin** carried out X-ray crystallography of DNA, which showed DNA to be a helix with even diameter

- **Watson and Crick model** of DNA: DNA is described as an **antiparallel double helix**
- Base pairing rule: A = T and G = C
- The nucleotides of the opposite chains are paired to one another by **hydrogen bonds**
- Hydrogen bonds are weak bonds that maintain the **spiral helix shape**

## DNA Duplication

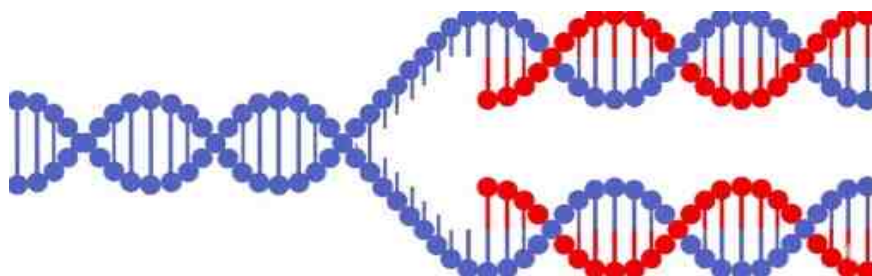
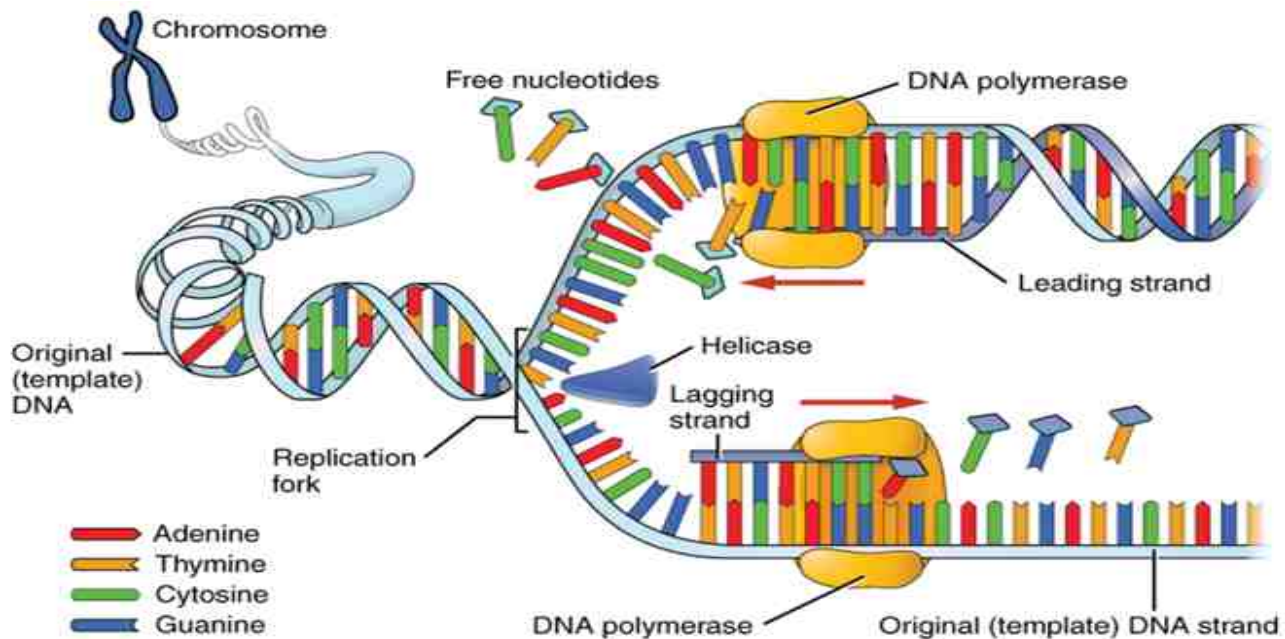
DNA duplication is the process of making an exact copy of DNA.

### Steps of Replication:

- 1- **Helicase enzyme** unwinds the double helix, breaks the hydrogen bonds, and separates the two strands.
- 2- **Each strand acts as a template** to make a complementary one.
- 3- **DNA polymerase** joins the new nucleotides together to make a new strand.

### Telomeres:

Telomeres are special non-coding nucleotide sequences at the end of chromosomes that are repeated thousands of times to **protect against possible loss of genes**.



Semi conservative replication

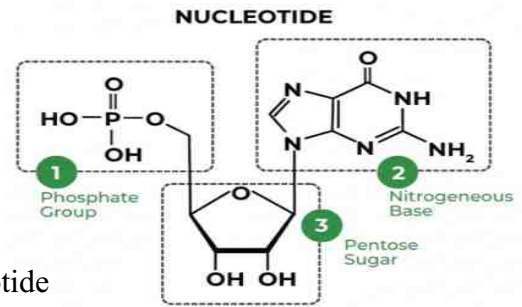
## 4. GENE EXPRESSION

Definition:

A gene is expressed when a certain protein is synthesized.

**Gene expression = protein synthesis.**

RNA is a polymer of ribonucleotides. The three parts of a nucleotide



**Genetic Codon:**

- A triplet of nucleotides carries a code for an amino acid.
- 1 gene = 1 protein.
- A base sequence on DNA determines the amino acid sequence on a protein.

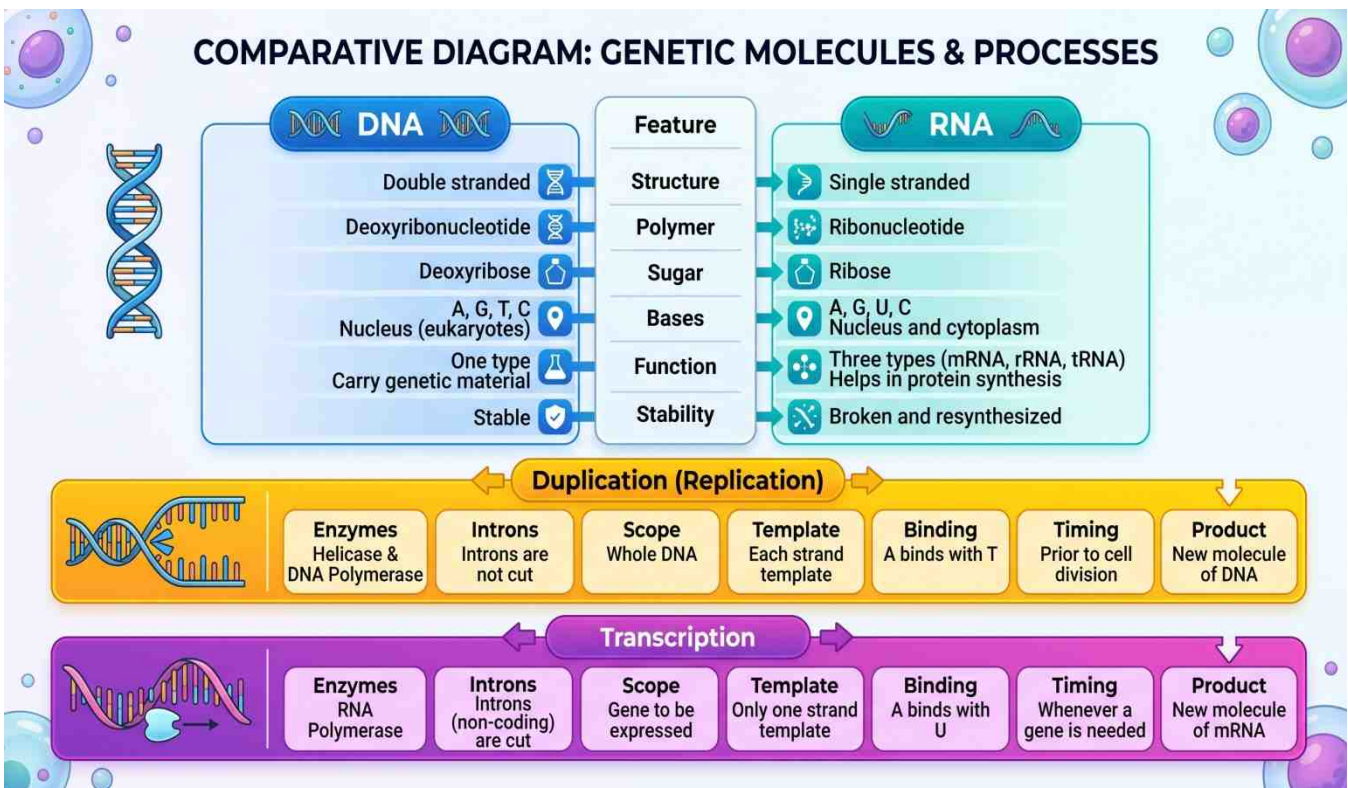
Notes:

- 64 codons and 20 amino acids.
- Each amino acid may be coded by more than one codon.

|              |   | Second letter                            |                                      |                                            |                                           |                  |
|--------------|---|------------------------------------------|--------------------------------------|--------------------------------------------|-------------------------------------------|------------------|
| First letter |   | U                                        | C                                    | A                                          | G                                         |                  |
|              | U | UUU } Phe<br>UUC }<br>UUA } Leu<br>UUG } | UCU } Ser<br>UCC }<br>UCA }<br>UCG } | UAU } Tyr<br>UAC }<br>UAA Stop<br>UAG Stop | UGU } Cys<br>UGC }<br>UGA Stop<br>UGG Trp | U<br>C<br>A<br>G |
| C            | C | CUU } Leu<br>CUC }<br>CUA }<br>CUG }     | CCU } Pro<br>CCC }<br>CCA }<br>CCG } | CAU } His<br>CAC }<br>CAA } Gln<br>CAG }   | CGU } Arg<br>CGC }<br>CGA }<br>CGG }      | U<br>C<br>A<br>G |
| A            | A | AUU } Ile<br>AUC }<br>AUA }<br>AUG Met   | ACU } Thr<br>ACC }<br>ACA }<br>ACG } | AAU } Asn<br>AAC }<br>AAA } Lys<br>AAG }   | AGU } Ser<br>AGC }<br>AGA } Arg<br>AGG }  | U<br>C<br>A<br>G |
| G            | G | GUU } Val<br>GUC }<br>GUA }<br>GUG }     | GCU } Ala<br>GCC }<br>GCA }<br>GCG } | GAU } Asp<br>GAC }<br>GAA } Glu<br>GAG }   | GGU } Gly<br>GGC }<br>GGA }<br>GGG }      | U<br>C<br>A<br>G |

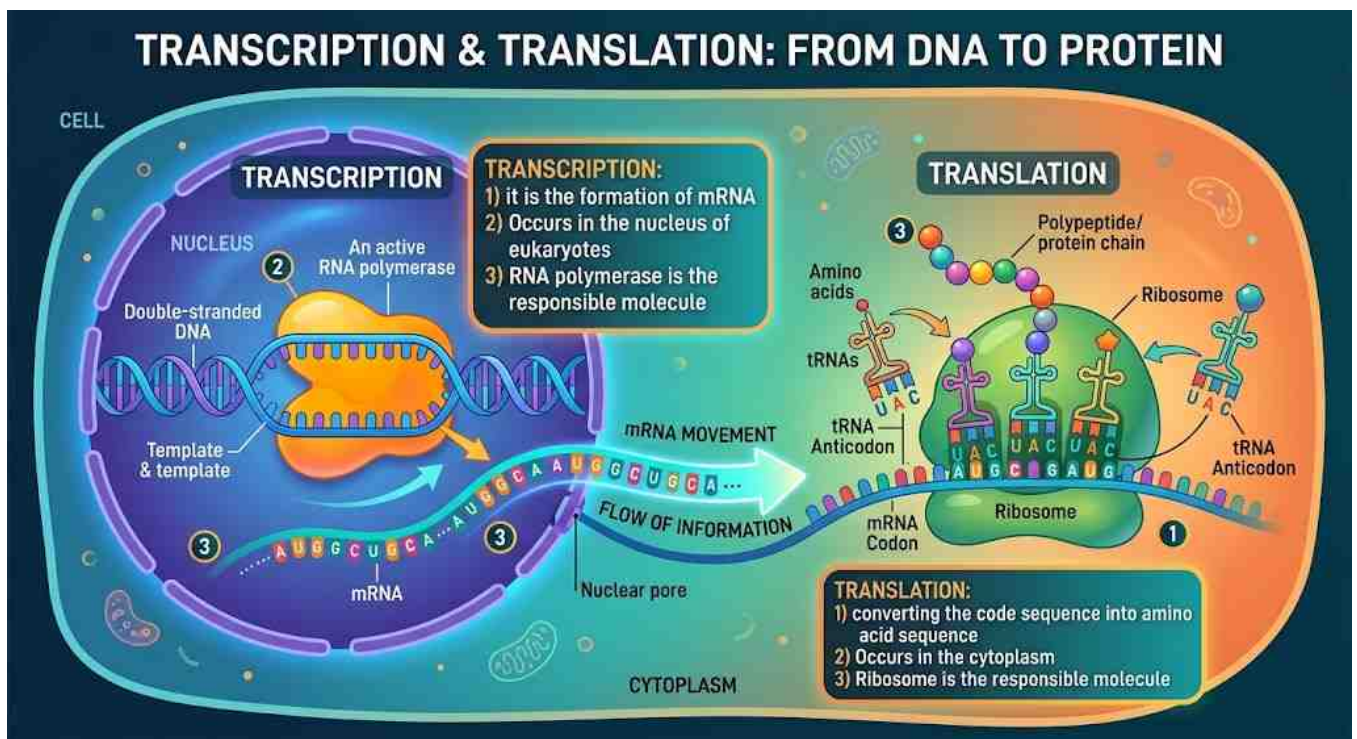
**Types of RNA:**

- **mRNA:** Transfers information from the nucleus (DNA) to the cytoplasm (ribosome).
- **rRNA:** rRNA and protein form the structure of the ribosome (the translator) that converts the base sequence to an amino acid sequence.
- **tRNA:** Brings the amino acids according to the codon on the mRNA.



- **Transcription:** formation of mRNA
- **RNA Processing:** introns “non-coding” are removed (RNA maturation)
- **Translation:** converting the code sequence into amino acid sequence
- If the protein consists of 50 amino acids, then there were 51 codons and 153 nucleotides
- tRNA has two sites: anticodon site and a.a attachment site
- **AUG** “start codon” while stop codon or End codon may be **UAA, UAG, UGA**

| Transcription                                 | Translation                                                |
|-----------------------------------------------|------------------------------------------------------------|
| 1) “it is the formation of mRNA”              | 1) “converting the code sequence into amino acid sequence” |
| 2) Occurs in the nucleus of eukaryotes        | 2) Occurs in the cytoplasm                                 |
| 3) RNA polymerase is the responsible molecule | 3) Ribosome is the responsible molecule                    |



## CONTROL OF GENE EXPRESSION: LAC OPERON IN E. COLI

- Operon is formed of promoter, operator, and functional (structural) genes
- The promoter is the binding site for RNA polymerase
- The operator is the binding site for the repressor
- TATA box (sequence of A and T) helps RNA polymerase bind to the promoter

### WITHOUT LACTOSE

- The repressor binds to the operator and blocks RNA polymerase from making mRNA for enzymes

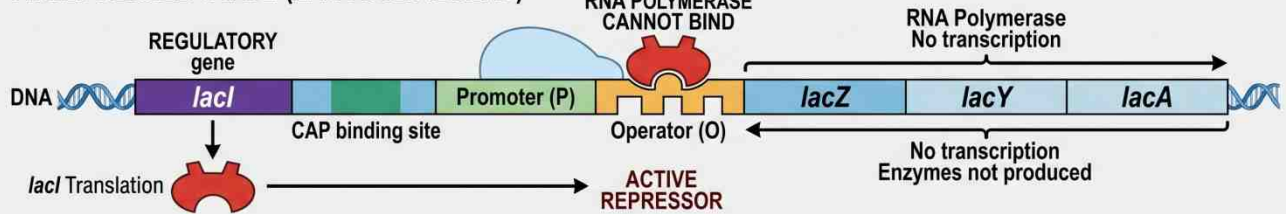
### WITH LACTOSE

- Lactose binds to the repressor and prevents it from binding to the operator
- RNA polymerase then synthesizes mRNA for structural enzymes

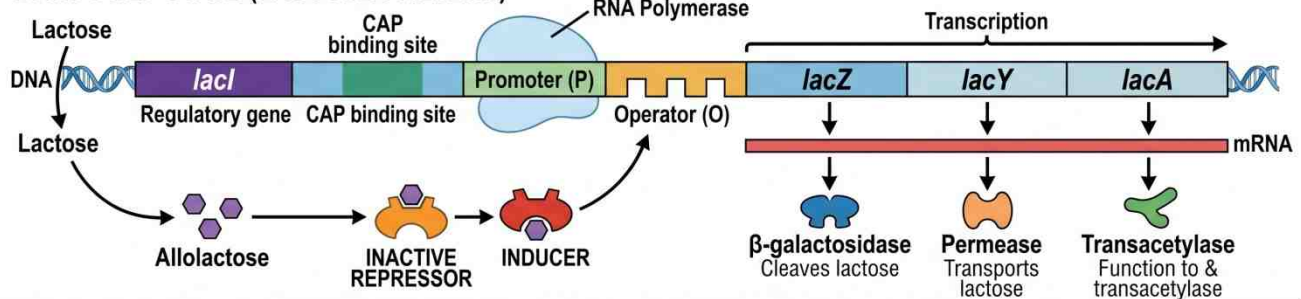
## CONTROL OF GENE EXPRESSION: LAC OPERON IN E. COLI

### NEGATIVELY REGULATED STATES

#### A. REPRESSED STATE (LACTOSE ABSENT)



#### B. INDUCED STATE (LACTOSE PRESENT)



## Gene Technology

### Human Genome

An organism's genetic material consists of about 3 billion base pairs of DNA and about **50,000 to 100,000 genes**. 97% of our DNA doesn't code for protein products; these are called junk DNA and include regulatory sequences and introns.

### Restriction Enzymes

Restriction enzymes cut DNA at specific recognition sites, such as GAATTC, and are sometimes referred to as **molecular scissors**. It comes from bacteria.

Applications:

- Recombinant DNA
- PCR
- Genetic fingerprinting

## Recombinant DNA

Recombinant DNA involves taking DNA from two sources and combining them into one.

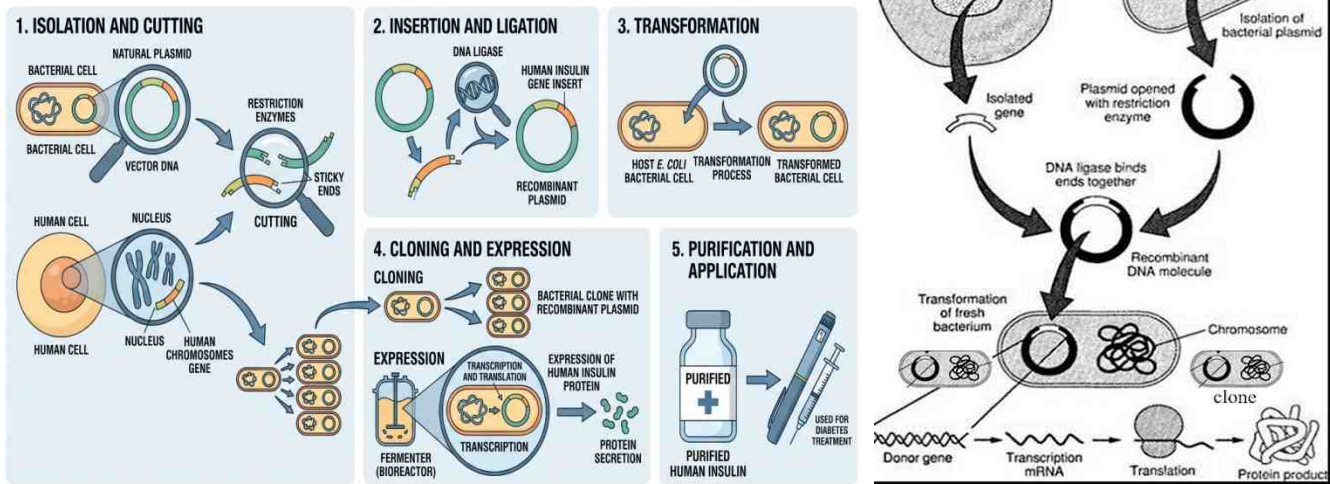
Example: Insulin DNA or any other Hormone or Enzyme

Using **Restriction enzymes** to cut DNA and **Ligase Enzyme** for joining it.

Notes:

- **Plasmids**: Ring-shaped DNA found in prokaryotes/bacteria.
- Apply the same restriction enzyme to both human and bacterial genetic material.
- The gene is isolated by **ultracentrifugation**.

### RECOMBINANT DNA (rDNA) TECHNOLOGY: PRODUCTION OF HUMAN INSULIN



### HOW POLYMERASE CHAIN REACTION (PCR) WORKS

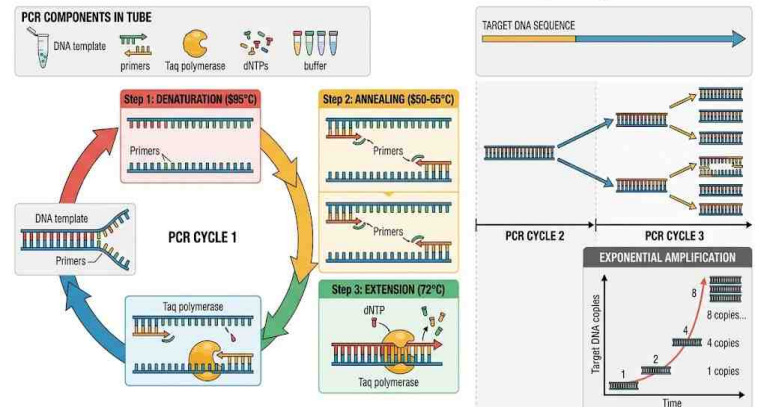
#### B. Polymerase Chain Reaction (PCR)

PCR is a cell-free, automated technique by which a piece of DNA can be rapidly copied or amplified.

Requirements:

- Original DNA
- Plenty of nucleotides
- DNA polymerase

Note: PCR is also described as **DNA amplification**



#### C. Genetic Fingerprint

There is a part of DNA known to be unique for each individual.

#### Gel Electrophoresis

Gel electrophoresis separates large molecules of DNA on the basis of their rate of movement through an agarose gel in an electric field.

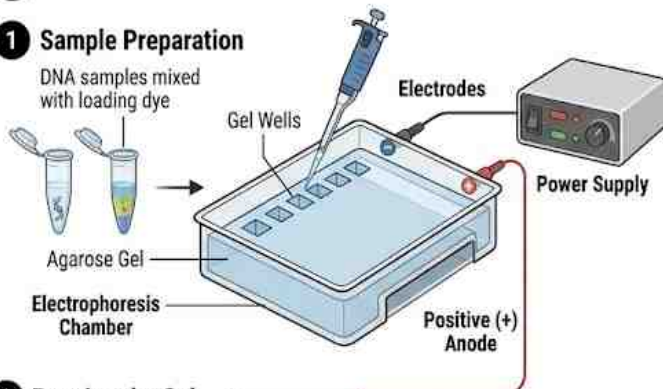
Key Principle:

- **The smaller the molecule, the faster it runs through the gel.**

## 1 PROCESS OF GEL ELECTROPHORESIS

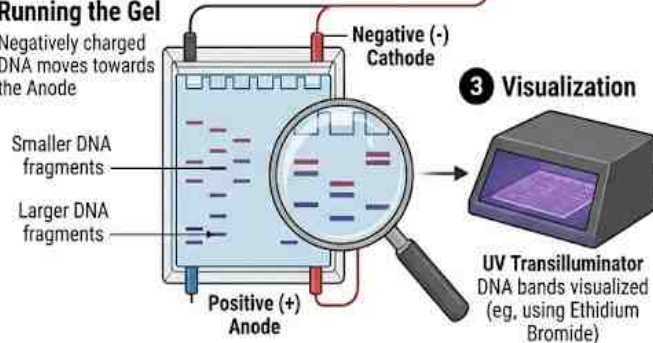
## 1 Sample Preparation

DNA samples mixed  
with loading dye

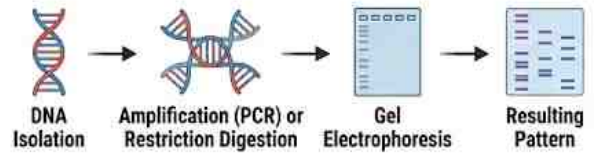


## 2 Running the Gel

Negatively charged DNA moves towards the Anode



## 2 APPLICATION: CREATING A GENETIC FINGERPRINT (DNA PROFILING)



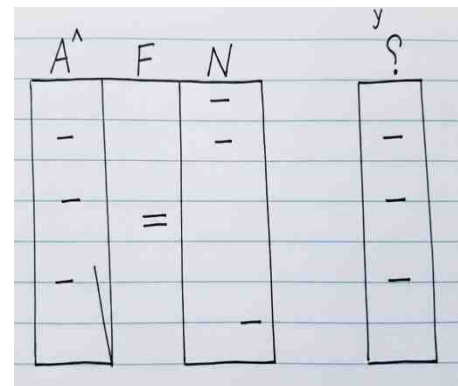
| COMPARISON FOR IDENTIFICATION |                                |           |           |                |         |          |
|-------------------------------|--------------------------------|-----------|-----------|----------------|---------|----------|
| DNA Ladder<br>(Size Marker)   | Evidence<br>(from Crime Scene) | Suspect A | Suspect B | Paternity Test |         |          |
|                               |                                |           |           | Mother 1       | Child 1 | Father 2 |
|                               |                                |           |           |                |         |          |
|                               |                                |           |           |                |         |          |
|                               |                                |           |           |                |         |          |
|                               |                                |           |           |                |         |          |
|                               |                                |           |           |                |         |          |
|                               |                                |           |           |                |         |          |
|                               |                                |           |           |                |         |          |

**Genetic Fingerprint:**  
A unique pattern of DNA bands specific to an individual

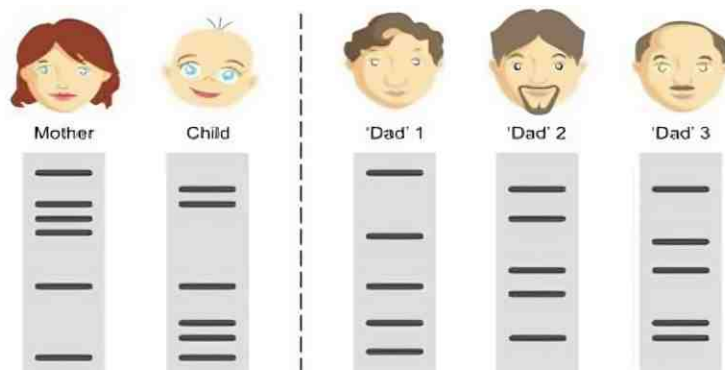
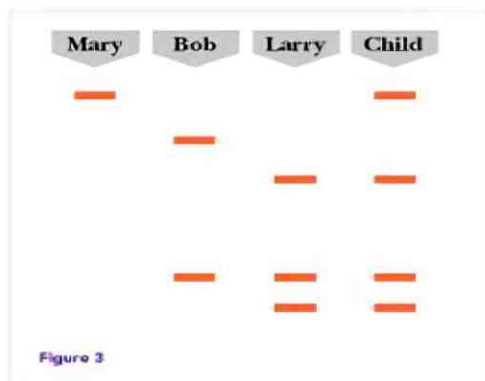
**MATCH:**  
Identifies Suspect A

### Case Study 1:

In a crime scene investigation, the police found a biological sample (hair, saliva, blood, or cells) in the nail of the victim. There are three suspects: Ahmed, Fares, and Nadine



## Case Study 2:



## Gene Mutation

Sudden changes in the genetic material or primary source of genetic variations.

Types of Mutations

- **Somatic** (Body cell): Example: cancer
- **Germ** (Gametes or gonads): Inherited
- **Gene** mutation
- **Chromosomal** mutation

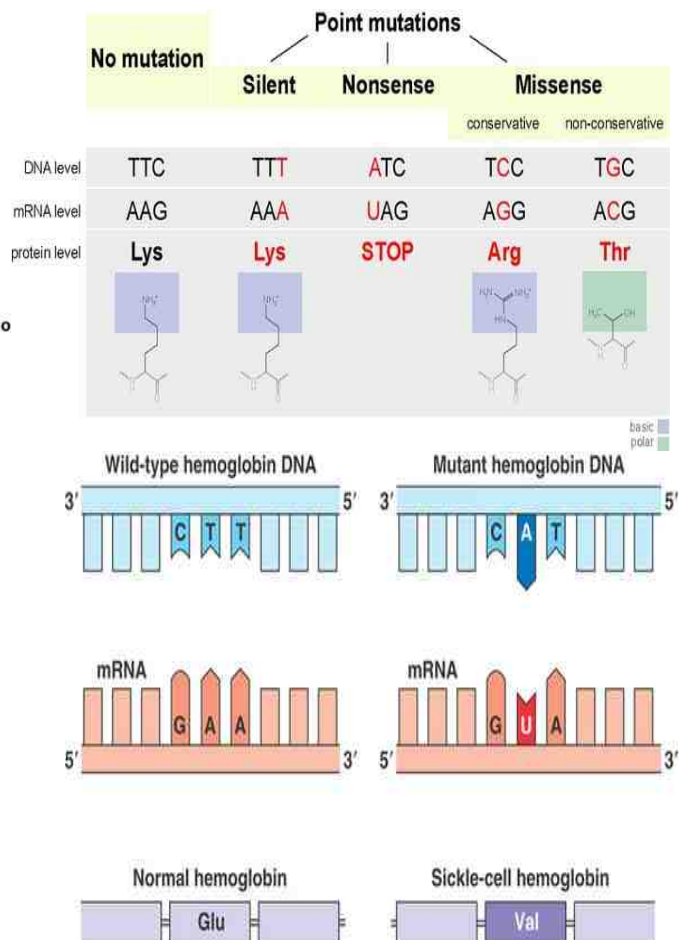
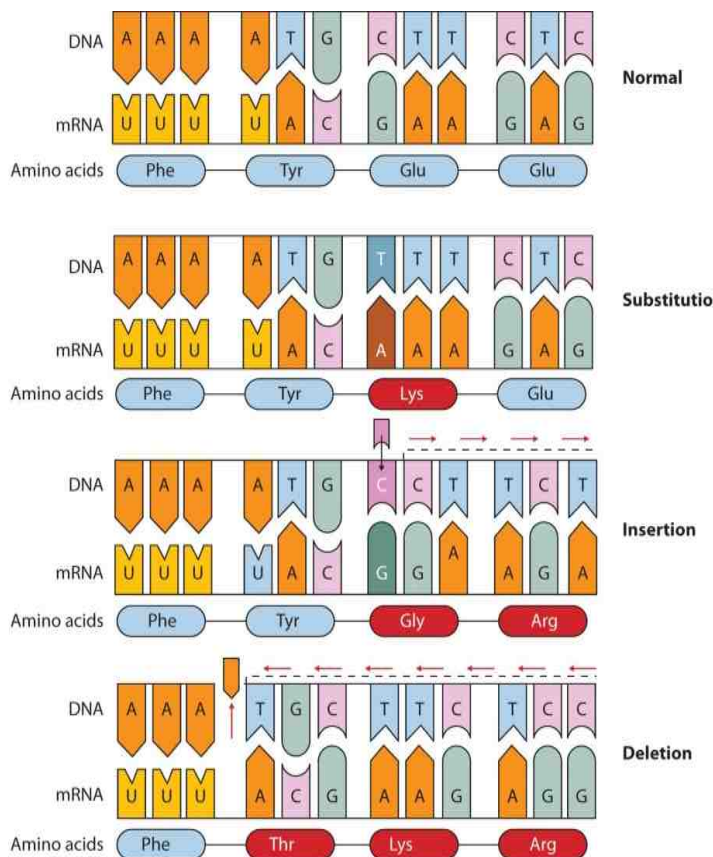
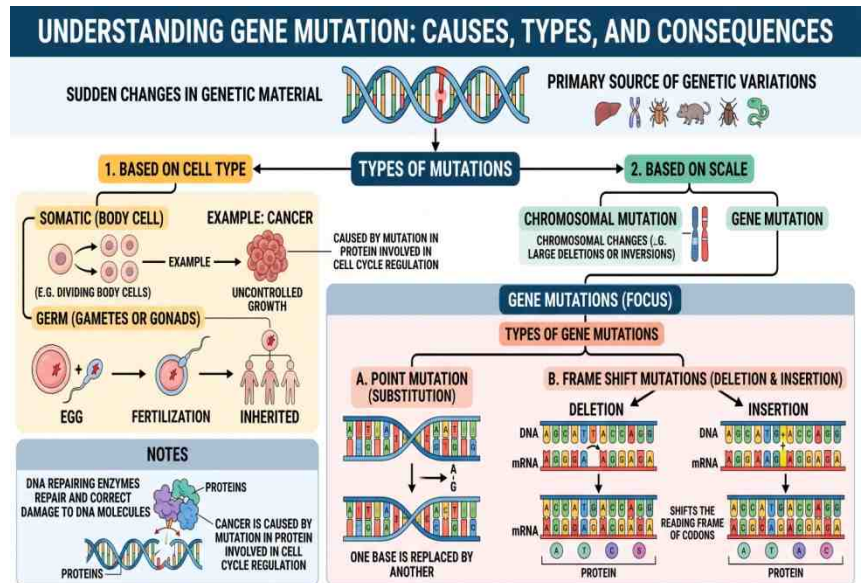
Notes

- DNA repairing enzymes repair and correct damage to DNA molecules
- Cancer is caused by a mutation in protein involved in cell cycle regulation

## Gene Mutation

Types of Mutations:

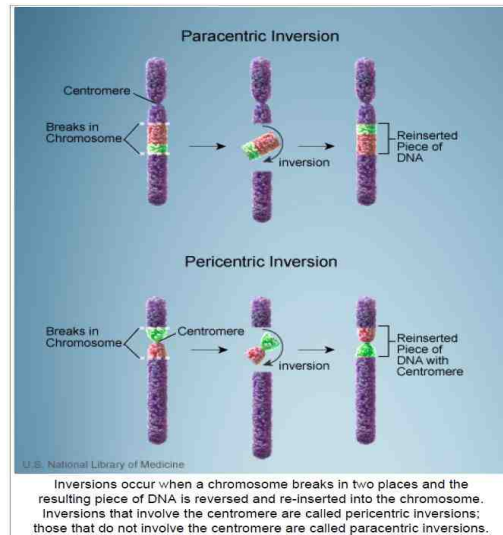
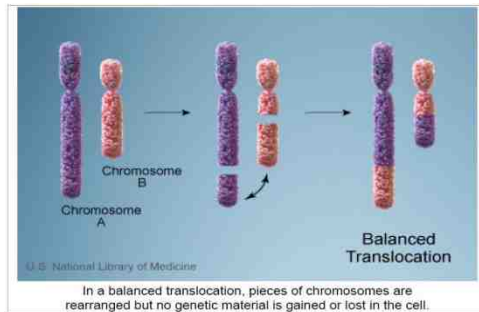
- **Point mutation (substitution)**
- **Frame shift mutations (Deletion and Insertion)**



## Chromosomal Mutations (Chromosomal Aberrations)

Types:

- **Inversion**
- **Translocation** (non homologous chromosomes)
- **Non-disjunction** (change in number)



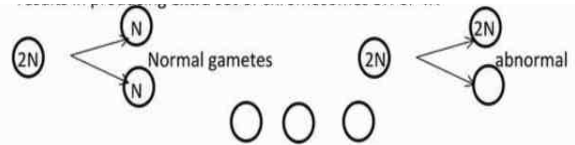
### Non-disjunction: Polyploidy and Polysomy

**Aneuploidy** is defined as having any abnormal number of chromosomes

**A-Polyploidy** is due to the inability of spindle fibers to separate the pairs of chromosomes, resulting in the production of extra chromosomes ( $3N$  or  $4N$ ).

Note:

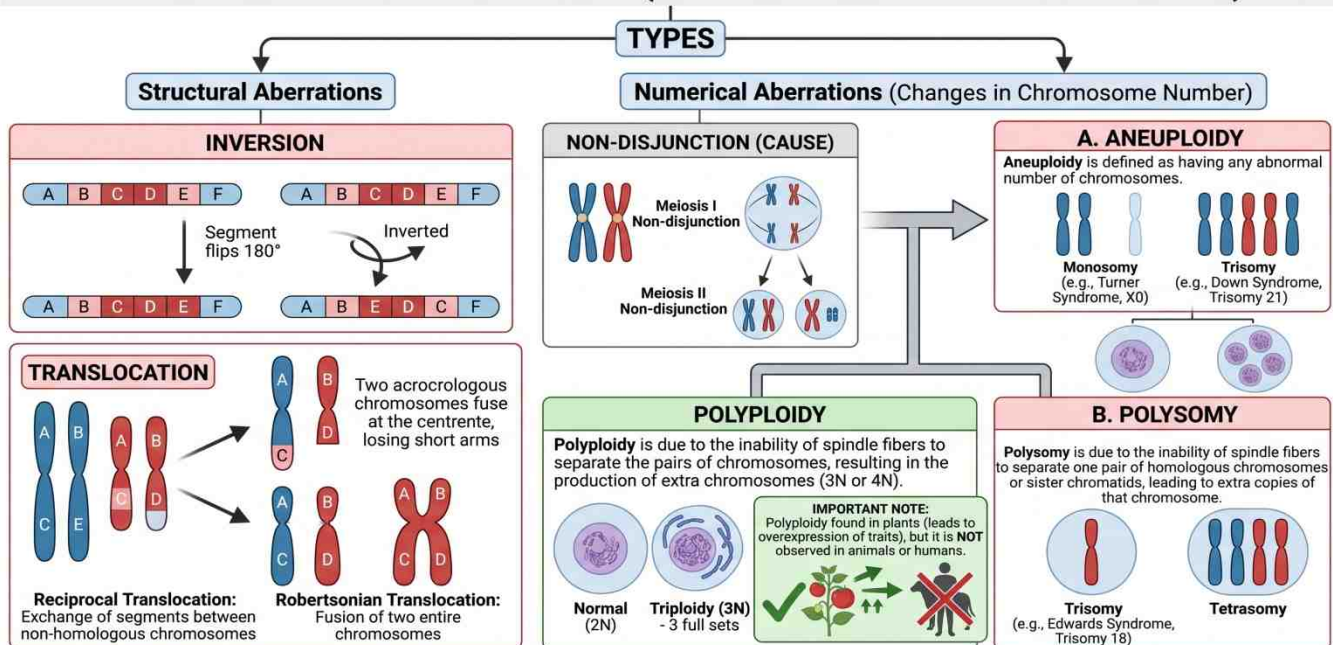
**Polyploidy is found in plants** and leads to overexpression of traits, but it is not observed in animals or humans.



### B. Polysomy

Polysomy is due to the inability of spindle fibers to separate **one pair of chromosomes**.

## CHROMOSOMAL MUTATIONS (CHROMOSOMAL ABERRATIONS)

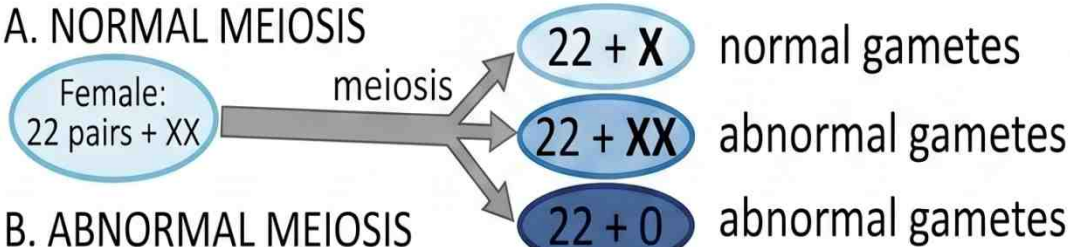


# CHROMOSOMAL DISORDERS IN HUMANS

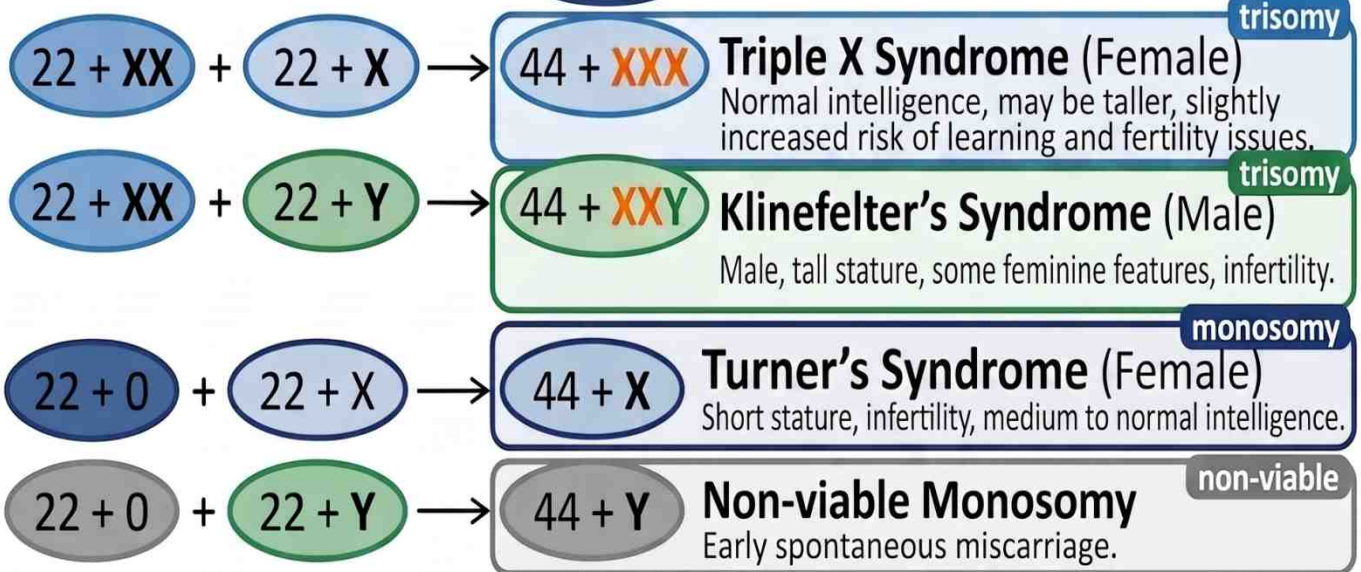
## اضطرابات الكروموسومات في الإنسان

### I. SEX CHROMOSOME ABNORMALITIES

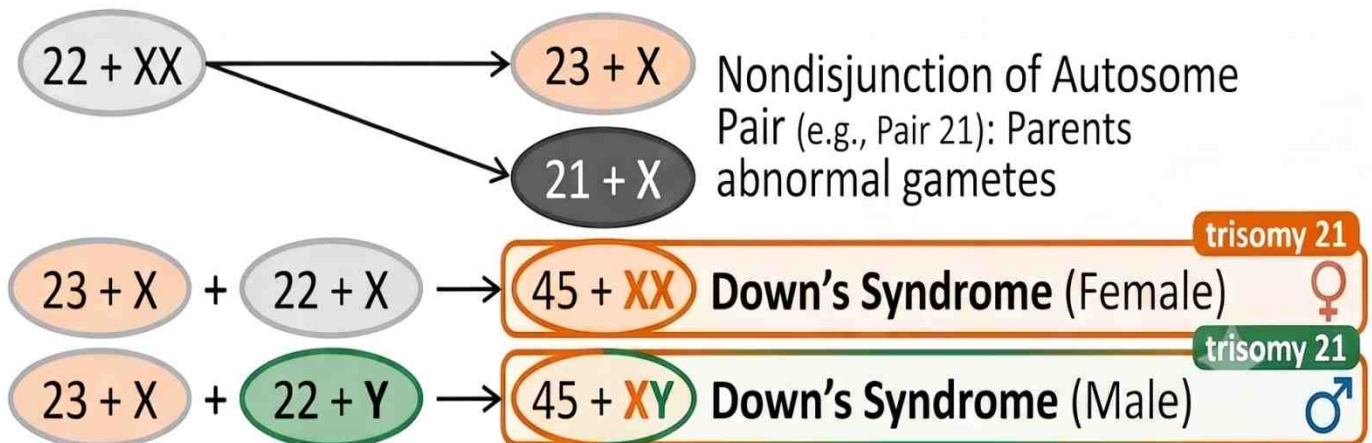
#### A. NORMAL MEIOSIS



#### B. ABNORMAL MEIOSIS



### II. AUTOSOMAL CHROMOSOME ABNORMALITIES (e.g., Down's Syndrome)



**Note:** Down's syndrome is a trisomy of chromosome pair 21.

# Diagram of Genetic Disorders and Their Patterns of Inheritance

## Patterns of Inheritance

### Autosomal Recessive

Recessive gene must be inherited from both parents.

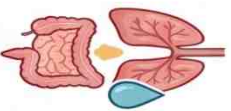
#### Phenylketonuria (PKU)

Requires phenylalanine-free diet to prevent mental retardation.



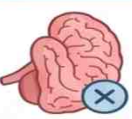
#### Cystic fibrosis

Buildup of thick extracellular fluid in lungs and digestive system.



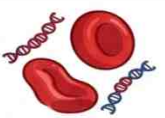
#### Tay-Sachs disease

Lack of enzyme to break down lipids in brain, to seizures & early death.



#### Thalassemia

Hereditary disease from faulty hemoglobin synthesis.

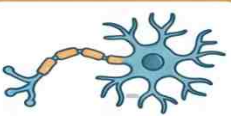


### Autosomal Dominant

Dominant gene inherited from at least one parent.

#### Huntington's disease

Degeneration of the nervous system; onset typically in middle age.



### Sex-linked Recessive

Recessive gene on X chromosome; typically affects males

#### Hemophilia

Lack of blood clotting proteins, causing excessive bleeding.



#### Color blindness

Inability to distinguish between red and green colors.

